MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,ARSAK	SA00047974	34	M	SA00047974
Ordering Phys				**DOB** 06/15/1978
Client Order # SA00047974	**Account Information**			**Report Notes**
Collected 07/26/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/29/2012 10:18				

Test	Flag	Results	Unit	Reference Value	Perform Site*
ARSA Gene, Known Mutation			REPORTED 08/02/2012 09:40		
Specimen		Blood			MCR
Specimen ID		1038438			MCR
Order Date		26 Jul 2012 08:50			MCR
Reason For Referral					MCR

Family history of metachromatic leukodystrophy (MLD). Test for the presence of familial alterations within the ARSA gene.

Method MCR

DNA sequence analysis was used to test for the presence of the c.459+1G>A alteration in intron 2 in the ARSA gene. Analysis for this specific alteration was performed because it was previously identified in a family member. Mutation nomenclature is based on GenBank accession number NM_000487.4.

Result MCR

The following heterozygous sequence change was detected:
Intron: 2
DNA change: c.459+1G>A
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

The presence of a mutation previously identified in a family member indicates that this individual is a carrier of arylsulfatase A deficiency. This interpretation assumes that this individual is healthy and is not clinically affected with metachromatic leukodystrophy.

Since a mutation has been identified in the ARSA gene, genetic testing of at risk family members is possible. If appropriate, enzymatic and molecular testing should be offered to this individual's reproductive partner to further clarify their risk of having a child with MLD.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test.

Reviewed By	Devin Oglesbee PhD	MCR
Release Date	02 Aug 2012 09:37	MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.